Merchant & Gould P.C.
Inventor: Gill, et al.
Docket No.: 7500.360US01
Title IMPROVEMENTS IN AND RELATING TO ANALYSIS OF DNA SAMPLES
Attorney Name John J Gresens
Phone No 612 371 5265
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Fig. 1

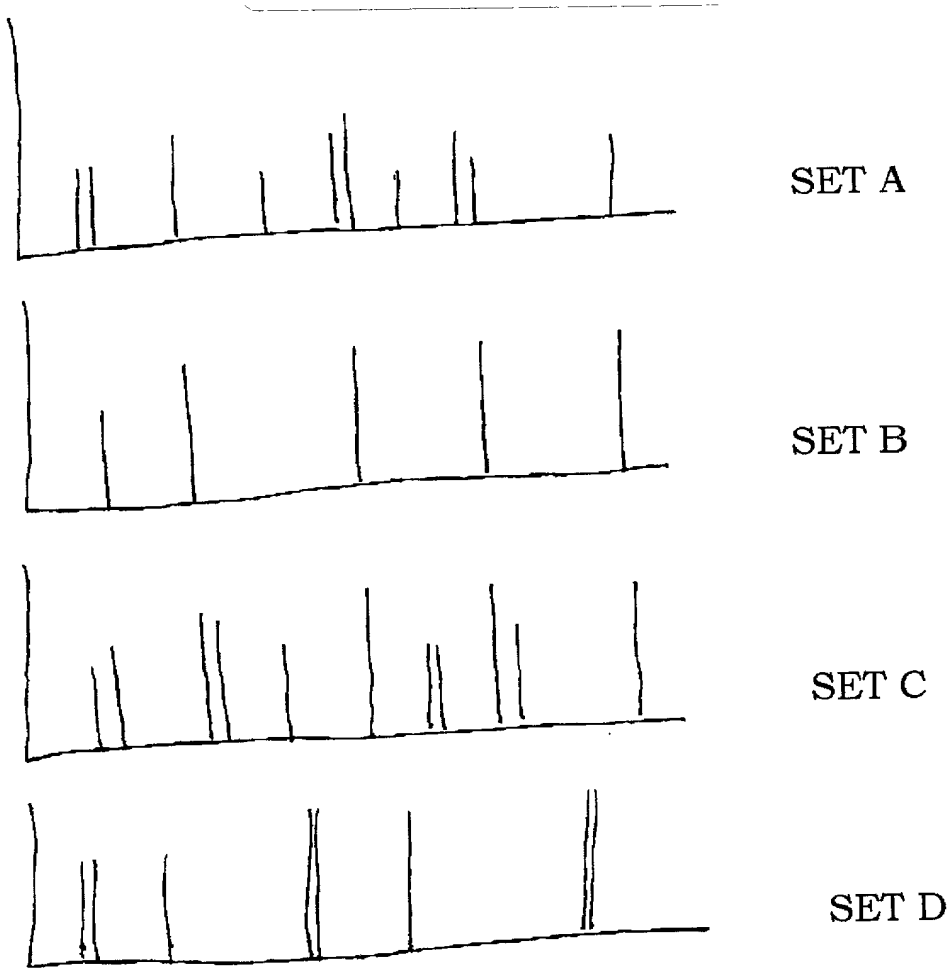
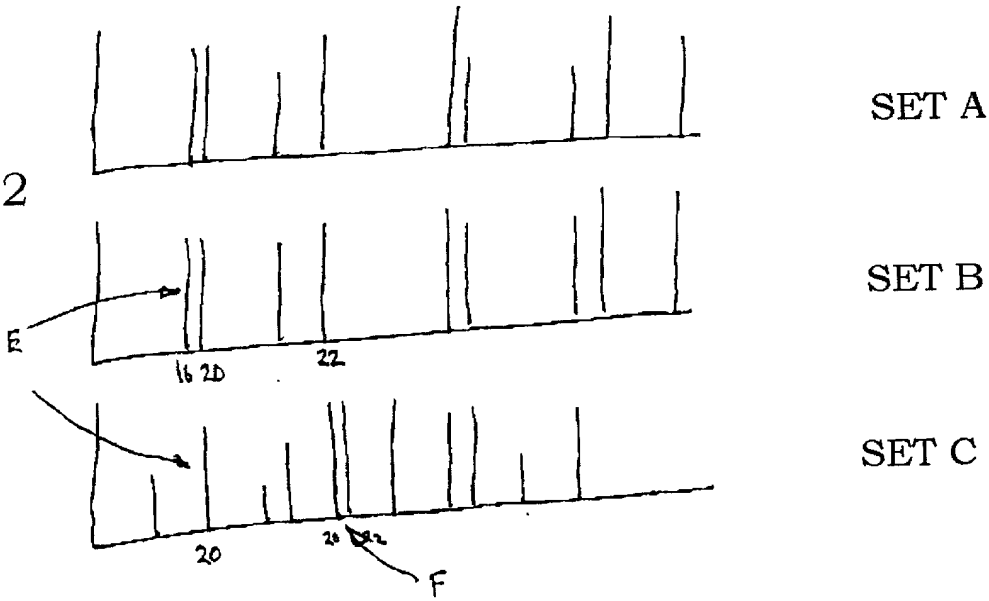


Fig. 2



TOETHO" 228h6360

M_j	$P(M_j)$	$R_1(12)$	$R_2(16)$	$R_3(12,16)$	Products
12 12	f_{12}^2	$p(\bar{D})p(\bar{C})$	$p(D)p(C)f_{16}$	$p(\bar{D})p(C)f_{16}$	$p(\bar{D})^2 p(\bar{C})p(D)p(C)^2 f_{12}^2 f_{16}^2$
12 16	$2f_{12}f_{16}$	$p(D)p(\bar{C})p(\bar{D})$	$p(D)p(\bar{C})p(\bar{D})$	$p(\bar{D})^2 p(\bar{C})$	$2p(\bar{D})^4 p(\bar{C})^3 p(D)^2 f_{12}f_{16}$
16 16	f_{16}^2	$p(\bar{D})p(C)f_{12}$	$p(\bar{D})p(\bar{C})$	$p(\bar{D})p(C)f_{12}$	$p(\bar{D})^2 p(\bar{C})p(D)p(C)^2 f_{12}^2 f_{16}^2$
					Denominator=sum of above

Table 1: Calculation of the components of the likelihood ratio for an example where 3 replicates show evidence of spurious bands and allele drop-out.

Fig. 3

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TOETHO" 228h:E860

M_J	$P(M_J)$	$R_1=abc$	$R_2=ac$	Products
ab	$2f_af_b$	$p(\overline{D})^2 p(C) f_c p(\overline{St})^2$	$p(D)p(\overline{D})p(C)f_a p(\overline{St})^2$	$2f_af_bf_c^2 p(\overline{D})^3 p(D)p(C)^2 p(\overline{St})^4$
ac	$2f_af_c$	$p(\overline{D})^2 p(\overline{St}) \left(p(St)p(\overline{C}) + p(\overline{St})p(C)f_b\right)$	$p(\overline{D})^2 p(C)p(St)^2$	$2f_af_c p(\overline{D})^4 p(\overline{C})p(\overline{St})^3 \left[p(St)p(\overline{C}) + p(\overline{St})p(C)f_b\right.$
bc	$2f_bf_c$	$p(\overline{D})^2 p(C) f_a p(\overline{St})$	$p(\overline{D})p(D)p(C)f_a p(\overline{St})^2$	$2f_a^2 f_b f_c p(\overline{D})^3 p(D)p(C)^2 p(\overline{St})^3$

Table 2: Derivation of equation 12. Note that $p(\overline{St})$ only appears once when $M_J=bc$ and $R_1=abc$ because b must be in part or wholly allelic.

Fig. 4

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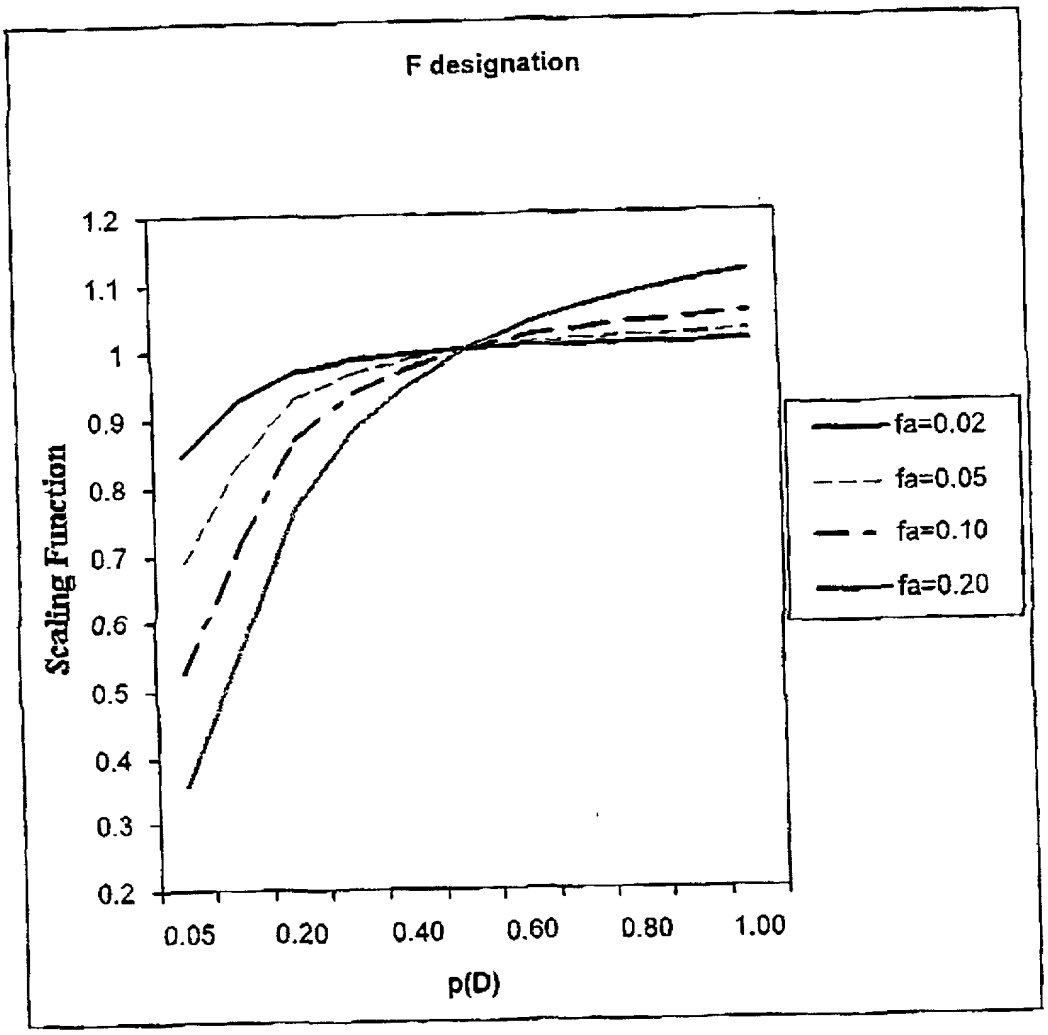
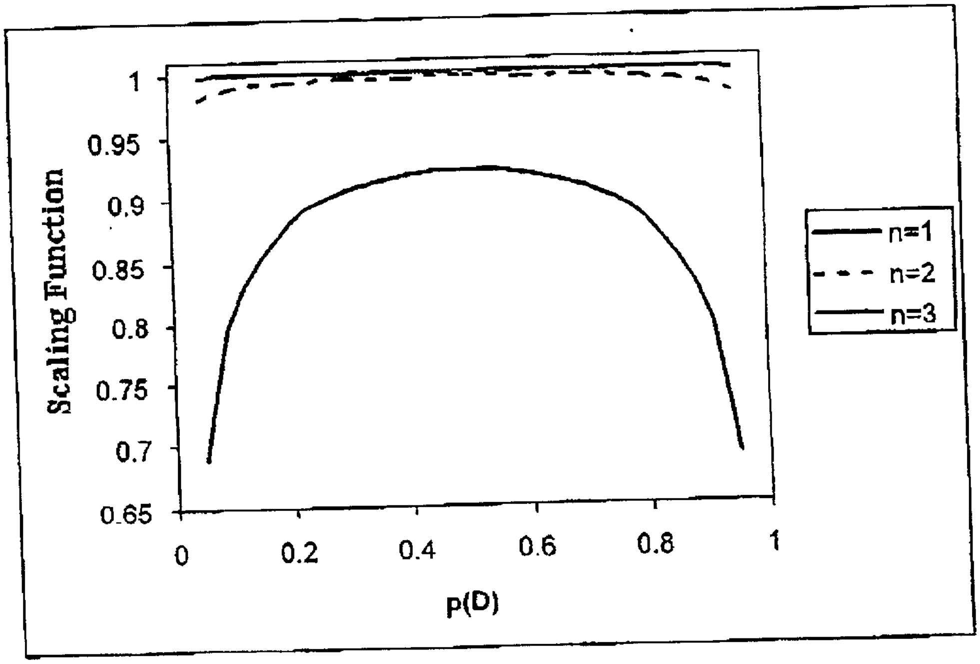


Testing the robustness of the F designation: The F designation is *conservative provided that the expression* $\frac{1}{\left[1+\frac{1-2p(D)}{2p(D)}f_a\right]}$ □1.0 (from equation 7).

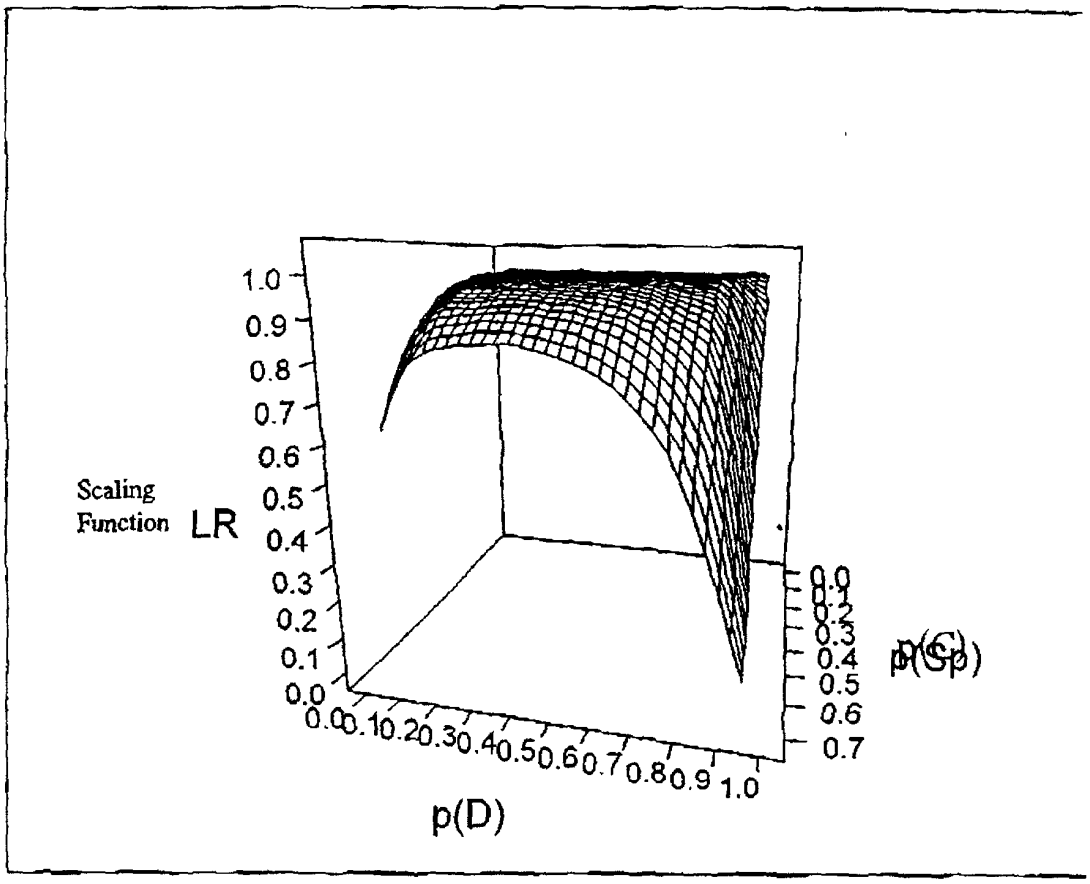
Allele frequencies (f_a) 0.02. 0.05, 0.10 and 0.20 are tested. Generally the F designation is conservative provided $p(D)$>0.5.

Fig. 5



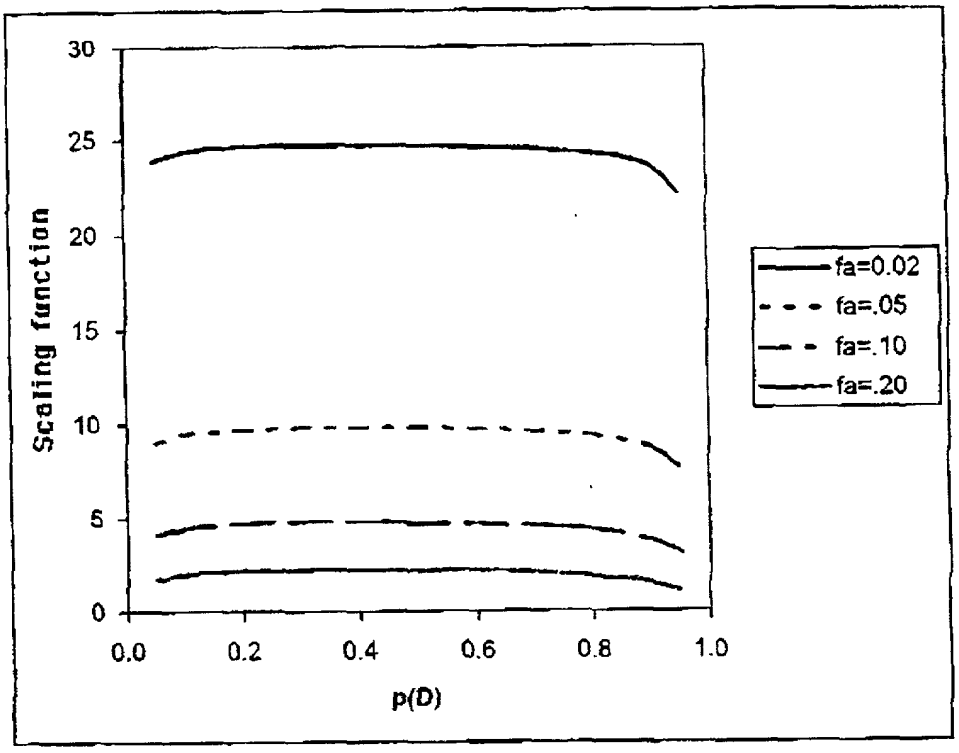
Evaluation of $\dfrac{1}{\left[1+\dfrac{f_a f_b^{n-1} p(C)^n}{2p(D)[p(D)p(C)]^n}\right]}$ from equation 9 (called the scaling function). When n (the number of replicates where the genotype is ab) is greater than or equal to 2 and $R_1 = a$, then the LR□$1/2f_af_b$. When n=1, $1/2f_a$ would be used which is always conservative. In fig 2a $p(C)$=0.3; f_a=f_b=0.1.

Fig. 6a



The 3 dimensional model showing the relationships between $p(C)$ and $p(D)$ when f_a=0.1. The scaling function □*1.0* for moderate and low values of $p(C)$ and for all intermediate values of p(D).

Fig. 6b

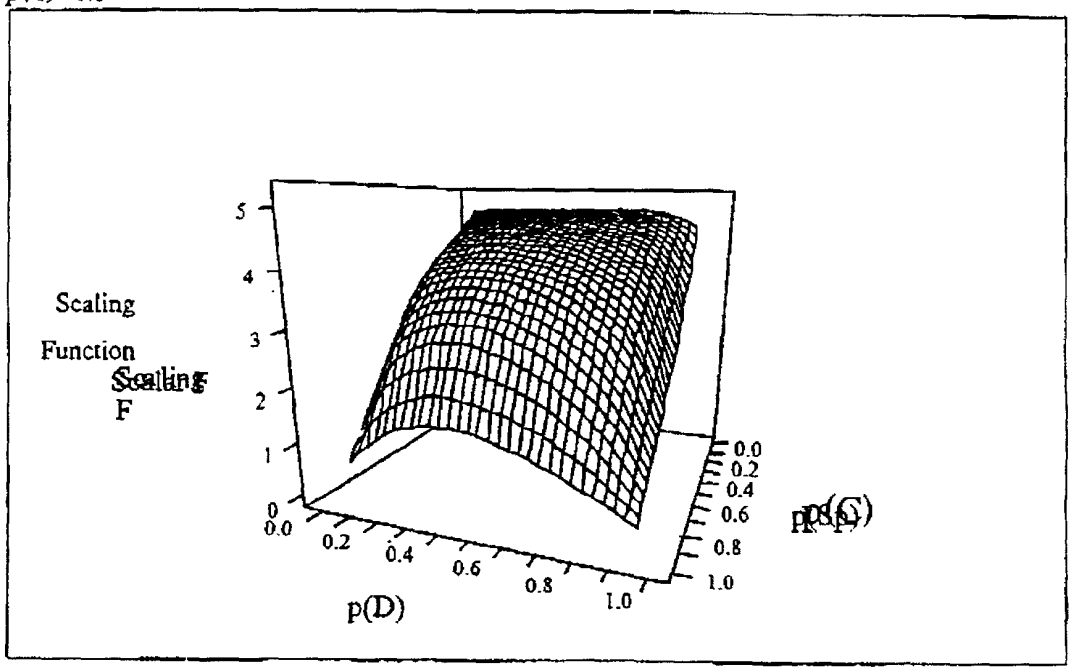


Provided that the scaling function $\frac{1}{f_a\left[2+\frac{f_b p(D)p(C)}{p(\bar{D})p(\bar{C})}+\frac{f_b p(C)}{2p(\bar{D})p(D)p(\bar{C})}\right]} \geq 1.0$

(from equation 10) then $1/2f_b$ is conservative. Allele frequencies are $f_a=f_b$=0.02,0.05,0.1,0.2 respectively and

Fig. 7a

p(C)=0.3.



3- dimensional graph to show the combined effect of *p(D)* and *p(C)* when f_a=0.10. The critical point where the scaling function >1 is reached when *p(C)*<0.9 and p(D)<0.9.

Fig. 7b

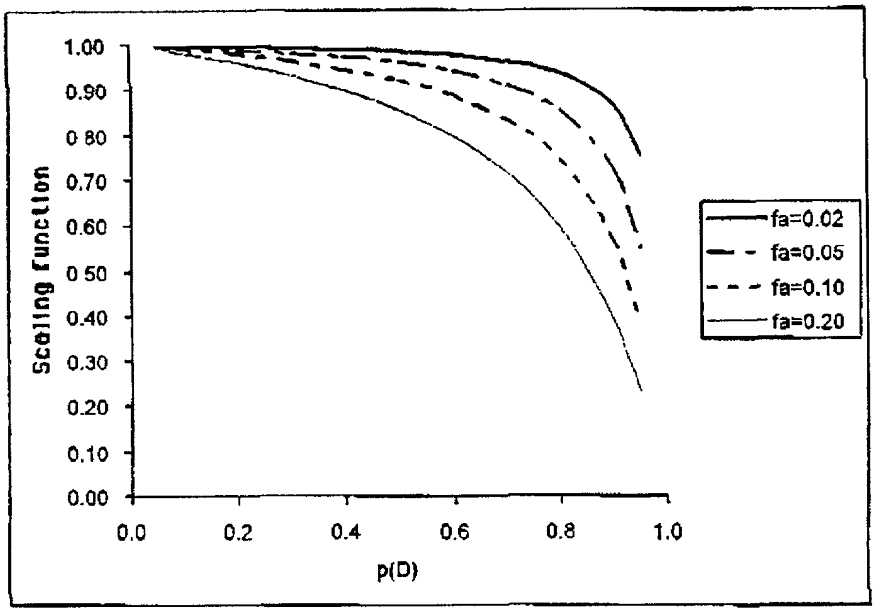
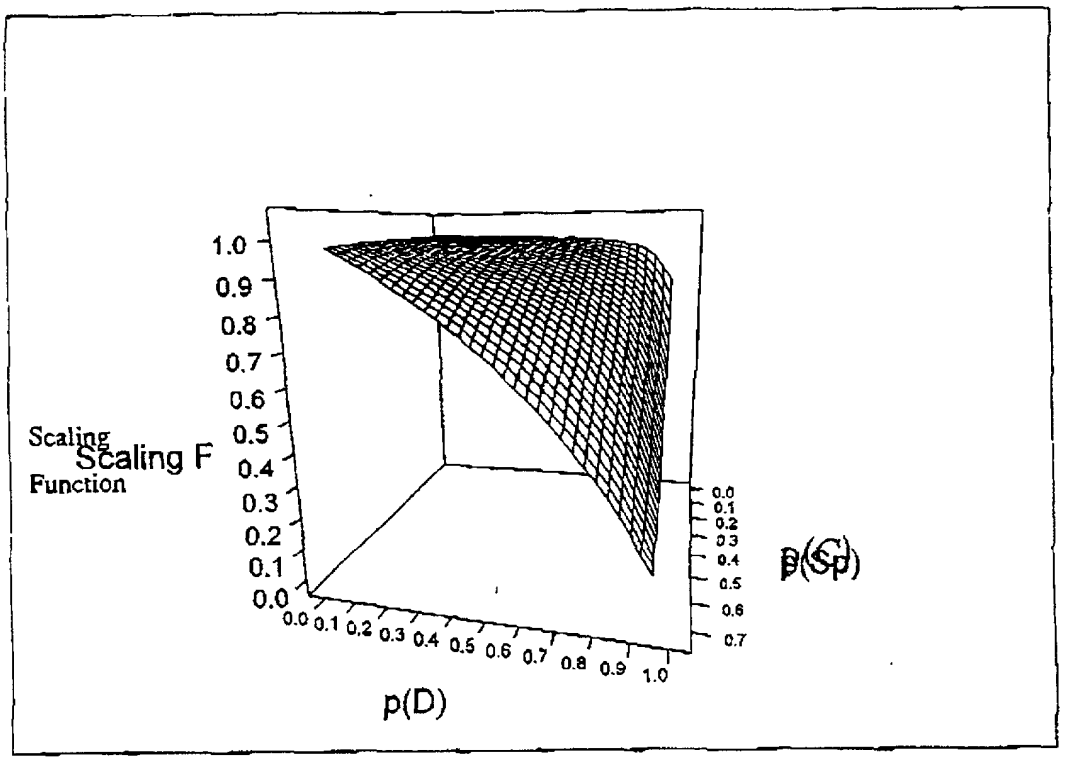
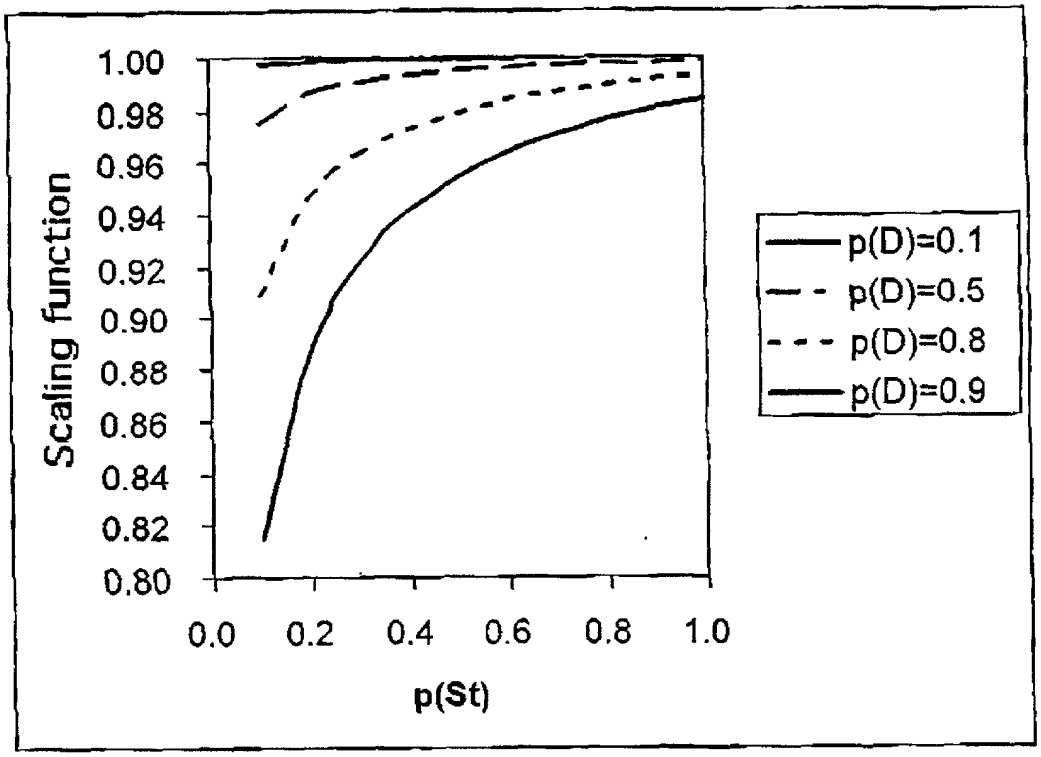

1.00
0.90
0.80
0.70
0.60
0.50
0.40
0.30
0.20
0.10
0.00
Scaling function
0.0
0.2
0.4
0.6
0.8
1.0
p(D)
fa=0.02
fa=0.05
fa=0.10
fa=0.20

Fig. 8a



When f_a=.1, the LR□1 when $p(D)$<0.5 and $p(C)$<0.3.

Fig. 8b



Plots of scaling function $\frac{1}{\left[1+\frac{p(C)^2 p(D) f_b (f_a + p(\overline{St}) f_c)}{p(\overline{C})p(\overline{D})\{p(St)p(\overline{C}) + p(\overline{St})p(C) f_b\}}\right]}$ (equation 12) v. $p(St)$ for several levels of $p(D)$ ranging from 0.1 – 0.9. $p(C)$=0.3 and fa=fb=fc=0.1.

Fig. 9

$p(D)$	Amelo	D19S433	D3S1358	D8S1179	HUMTH01	HUMVWFA31/A	D21S11	HUMFIBRA (FGA)	D16S539	D18S51	D2S1338	Mean
$p(D_{Ho})$	0.40	0.60	0.60	0.56		0.62						0.64
$p(D_{He})$					0.32		0.38	0.26	0.44	0.32	0.20	0.32
$p(D_{Hez})$					0.64		0.62	0.66	0.46	0.56	0.70	0.58

Table 3: Analysis of *p(D)* parameters derived from Table 1 of Gill et al [11].

Fig. 10